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A unique case of neurofibromatosis 1 (NF1) with Cotard syndrome – A case report

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Abstract

Neurofibromatosis type 1 is a neuro-cutaneous autosomal dominant disorder that is often associated with various psychiatric disorders. Cotard syndrome is a rare neuropsychiatric disorder seen in a variety of neurological and psychological illnesses. The complex course of the neurofibromatosis predisposes the individual towards higher physical and psychiatric morbidity. Here we present a case of Cotard syndrome in a patient with neurofibromatosis. The patient was brought with an alleged history of self-harm and pervasive sadness of mood; a diagnosis of recurrent depressive disorder current episode severe with psychotic features was made. Patient was treated with oral psychotropic agents and responded well to the treatment.

neurofibromatosis 1 (NF1); depression; Cotard Syndrome

INTRODUCTION:

Neurofibromatosis 1 (NF1) is an autosomal dominant disorder which affects approximately 1 in 3000 people [1]. Phenotypic manifestations noted in NF1 are café-au-lait spots, skinfold freckling, iris hamartomas, lisch nodules, bony dysplasias, and neurofibromas. They are predisposed to develop benign and malignant tumours [1,2]. It is also known to be associated with Autism, depression, dysthymia, anxiety and personality disorders [3,4]. Due to the cutaneous manifestations and its progressive course, the illness adversely affects the quality of life; which in turn leads to an increase in the psychological burden [5].

Amongst the psychiatric co-morbidities depression is the most common. The underlying factors theorized to be responsible for this are the visible cutaneous lesions and medical problems that put the individual at risk for societal stigmatization and discrimination. Prevalence of psychiatric co-morbidities is as high as 33%. The risk of suicide is increased by up to four times as compared to general population. Here we present a rare & unique case of Cotard syndrome, depression with NF1.

CASE REPORT:

A 30 years old male Mr. KB was brought by his father to outpatient services with a history of attempted self-harm by hanging. The patient reported feeling pervasive sadness of mood, inability to think or feel anything, hopelessness,

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loss of energy and suicidal ideations. The total duration of symptoms was five months. Patient had been unable to do any work since the onset of the low mood. During this time he would often wander on the streets apologizing to people and kept verbalizing his problems. Some nights he would not return to his home and would be found sleeping at the roadside.

In past, he had a prior episode of depression eight years back. During the episode he was not able to work due to low mood, would remain irritable and agitated and would wander in the village. He also had attempted self-harm by use of insecticide. After this the family consulted a mental health professional and he was started on oral medications. The details of the same are not available. The family discontinued the treatment due to financial difficulties.

No family history of psychiatric illness was elicited. Physical examination showed multiple neurofibromas with widespread distribution over the face, chest and back. He also had few café au lait spots the largest one measuring 26mm over the axilla.

On Mental status examination patient had poor grooming and hygiene. He reported his mood to be pervasively sad. His affect was appropriate to his mood with decreased range and reactivity. During interview he kept repeating that his body had rotten and he linked his bodily appearance to that of a dog; inferred as nihilistic delusions. He would feel that people kept staring at him. He also felt hopeless, helpless & worthless due to his condition. He felt that suicide was the only option which could end his suffering. Patient also reported elementary hallucinations of dogs barking and crows cawing. Based on the above he was diagnosed as recurrent depressive disorder, current episode severe with psychotic features.

In view of the suicidal attempt and active suicidal ideations, patient was admitted. All the routine blood investigations viz. haemogram, Renal function test, Liver Function test, Random Blood sugar, Thyroid profile, Vitamin B12 & Electrocardiogram were normal. Opinion from Neurology colleagues was sought for his NF1 and to rule out any organic pathology. MRI Brain Plain was done as part of evaluation and was found to be normal. ECT was offered as a treatment option in view of the depression and active suicidal ideation but the patient & relatives refused. He was started on Tab. Escitalopram 10 mg for depressive symptoms, Tab. Olanzapine 5 mg HS for psychotic features and T. Clonazepam 0.5 HS for sleep. Patient tolerated the medication and showed response hence the dosages were gradually built. Both Tab. Escitalopram and Tab. Olanzapine were increased to 20mg. After increasing the dose of medication within 2-3 weeks patient reported marked improvement in his mood and psychotic features; Clonazepam was discontinued as sleep was assured with the above regimen. Patient was maintained on the above treatment on out-patient basis.

DISCUSSION

The neurofibromatosis NF1, NF2, and schwannomatosis; are most common nerve sheath tumours [6]. Our patient had two episodes of depression with over a period of 8 years. The present episode was suggestive of severe depression, with psychotic features & Cotard syndrome.

In NF1 the psychological burden is likely to be high, but research regarding the potential psychological impact of NF in adulthood has been generally restricted to a few studies that have examined quality of life among patients with NF16–8 or NF29 and to a few that have assessed psychiatric morbidity among patients with NF1 only [6,7]. Also the exact etiopathology that can explain the co-occurrence of neurofibromatosis and psychosis are not clearly documented in the literature. What we know through genetic studies is the implication of mutations in learning disorders and certain cognitive functions. This is probably mediated through Ras-GAP activity of NF1 [8, 9]

However the genetic pathways, if any, responsible for psychiatric disorders had not yet been elucidated [10]. NF1 has been linked to high likelihood of depression and other psychiatric presentations [3, 11] NF1 affliction modifies the QoL, lowers the self-esteem and increases the perceived stress for the patients [3,5,6]. Effective management of this depression can improve the Qol [3].

Factors that could play a role in emotional and psychological problems seen in in NF1

- Potential stigmatization due to visible effects of the cutaneous lesions [11, 12, 13, 14].
- Living with NF and negative emotional consequences [6].
- Intellectual and social problems causing difficultly in adjusting
- Course and morbidity and complex nature of disease [2].

Management of psychiatric co-morbidity is focused on use of therapy, improving the family support and medications. We did find one case report of ECT use in neurofibromatosis patient (The patient also had other medical problems intracranial dermoid cyst and meningioma) [15].

Coming to the features of Cotard syndrome, it is a rare neuropsychiatric presentation with various psychopathology features ranging from nihilism to immortality delusions [16, 17]. The one stage experimental model explains it as anomalous perceptual experiences with global disconnection of sensory areas and limbic system [18]. Neuroanatomical correlates for the cause of the phenomenon have been lacking [16, 19]. An extensive list of psychiatric illnesses (viz. anxiety, depression, mental retardation, post-ictal depression etc.) and neurological disorders (viz. syphilis, migraine, epilepsy, brain tumours, brain injury etc.) have been seen in patients with Cotard syndrome [17, 20]. Management is by use of anti-psychotics, ECT or a combination of both. We however did not find any literature on Cotard syndrome in a case of neurofibromatosis 1.

CONCLUSION

In our case pharmacotherapy was effective. NF1 is currently devoid of an absolute cure and the individual has to lead a life with significant morbidity. Presence of Cotard syndrome and other such severe illnesses multiplies it many fold. Current guidelines for the management of NF1 patients focuses primarily on the medical aspects of the condition with only a brief mention of psychological problems. Regular screening for early identification of psychological co-morbidities and their treatment will help alleviate some burden in NF1.

Declaration of interest
The authors report no conflicts of interest.

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